

Suggested Amendments to Chapter 246-650 WAC, Newborn Screening

Submitted by Board Member Tom Locke, MD, MPH, October 15, 2003

The following are suggested amendments to proposed rules published as WSR 03-17-092.

1. Changes to the definition of galactosemia

(a) In proposed WAC 246-650-010(6), following the words “build up of galactose” add the words “and galactose-1-PO₄”.

(b) In proposed WAC 246-650-010(6), following the word “accumulated” change “galactose” to “galactose-1-PO₄”.

Effect: The changes would make the definition more technically accurate. Both compounds accumulate and the phosphorylated version, galactose-1-PO₄, causes the tissue and organ damage. C. Ronald Scott, MD, a Newborn Screening Advisory Committee member, suggested the amendments during the public comment period. If the Board adopts the suggested changes, the definition of galactosemia would read:

“Galactosemia” means a deficiency of enzymes that help the body convert the simple sugar galactose into glucose resulting in a buildup of galactose and galactose-1-PO₄ in the blood. If undetected and untreated, accumulated galactose-1-PO₄ may cause significant tissue and organ damage often leading to sepsis and death.”

2. Changes to the definition of homosystinuria

(a) In proposed WAC 246-650-010(7), following the words “enzymes necessary to” replace “breakdown” with “break down or recycle”.

(b) In proposed WAC 246-650-010(7), following the words “amino acid” replace “methionine” with “homocysteine”.

(c) In proposed WAC 246-650-010(7), following the words “buildup of methionine” replace “in the blood and buildup of homocystine in the urine” with “and homocysteine”.

Effect: The changes would make the definition be more concise and technically accurate since enzymes affecting both processes may be involved, homocysteine is the substrate, and the patterns of buildup vary with different forms of the disease. The amendments would also correct a grammatical error (“break down” in the verb form is two words). Dr. Scott suggested the amendments. If the Board adopts the suggested changes, the definition of hemocystinuria would read:

“Homocystinuria” means deficiency of enzymes necessary to break down or recycle the amino acid homocysteine resulting in a buildup of methionine and homocysteine. If undetected and untreated may cause thromboembolism, mental and physical disabilities.

3. Change to the definition of phenylketonuria

(a) In proposed WAC 246-650-010(12), following the words “phenylalanine in the blood” delete the words “and excretion of phenylketones in the urine”.

Effect: The change would make the definition more concise and technically accurate. Identifying the excretion products (which include phenylalanine metabolites phenylpyruvic acid, phenyllactic acid and phenylacetic acid as well as phenylalanine itself) is not necessary for the definition. Dr. Scott suggested the amendment during the public comment period. If the Board adopts the suggested change, the definition of phenylketonuria would read:

"Phenylketonuria" (PKU) means a deficiency of an enzyme necessary to convert the amino acid phenylalanine into tyrosine resulting in a buildup of phenylalanine in the blood. If undetected and untreated may cause severely impaired mental functioning.

4. Changes to the privacy and security provisions

(a) In proposed WAC 246-650-050(4), following the words "may only be released" insert the words "when required by state or federal law or".

Effect: The effect of the change would be to recognize that state and federal laws such as the Health Care Information Act might require the release of specimens and related information. If the Board adopts the suggested change, WAC 246-650-050(4) would read in part:

Release: Dried blood spot samples and specimen information may only be released when required by state or federal law or under the following conditions...